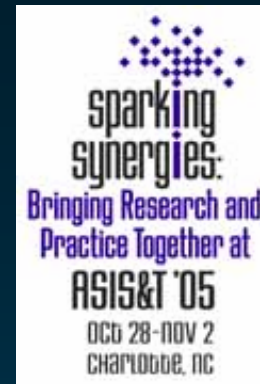


Session 33
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*Ontological research
and its applications to the biomedical domain*

Biomedical resources for text mining



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Overview

- ◆ An example
- ◆ Three types of resources
 - Lexical resources
 - Terminological resources
 - Ontological resources
- ◆ Some issues

An example

Neurofibromatosis 2

Neurofibromatosis 2

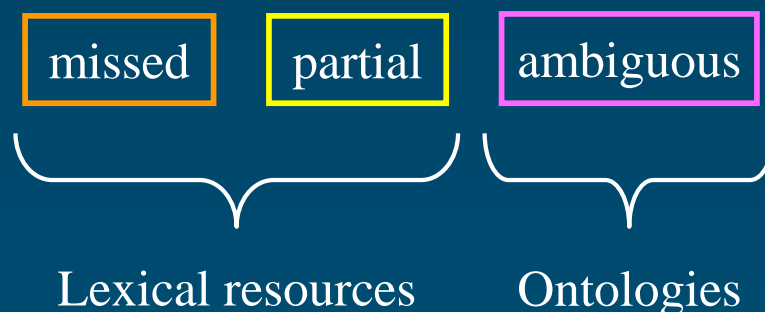
Neurofibromatosis type 2 (NF2) is often not recognised as a distinct entity from peripheral neurofibromatosis. NF2 is a predominantly intracranial condition whose hallmark is bilateral vestibular schwannomas. NF2 results from a mutation in the gene named merlin, located on chromosome 22.

[Uppal, S., and A. P. Coatesworth. "Neurofibromatosis Type 2." *Int J Clin Pract*, 57, no. 8, 2003, pp. 698-703.]



Entity recognition

Neurofibromatosis type 2 (NF2) is often not recognised as a distinct entity from peripheral neurofibromatosis. NF2 is a predominantly intracranial condition whose hallmark is bilateral vestibular schwannomas. NF2 results from a mutation in the gene named merlin, located on chromosome 22.



Relation extraction

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- vestibular schwannomas *manifestation of* neurofibromatosis 2
- neurofibromatosis 2 *associated with* mutation of NF2 gene
- NF2 gene *located on* chromosome 22

Resources for text mining

Types of resources

◆ Lexical resources

- Collections of lexical items
- Additional information
 - Part of speech
 - Spelling variants
- Useful for entity recognition
- UMLS SPECIALIST Lexicon, WordNet

◆ Ontological resources

- Collections of
 - kinds of entities (substances, qualities, processes)
 - relations among them
- Useful for **relation extraction**
- UMLS Semantic Network, SNOMED CT

Types of resources (revisited)

- ◆ Lexical and terminological resources
 - Mostly collections of names for biomedical entities
 - Often have some kind of hierarchical organization (e.g., relations)
- ◆ Ontological resources
 - Mostly collections of relations among biomedical entities
 - Sometimes also collect names

Unified Medical Language System



◆ SPECIALIST Lexicon

- 200,000 lexical items
- Part of speech and variant information

◆ Metathesaurus

- 5M names from over 100 terminologies
- 1M concepts
- 16M relations

◆ Semantic Network

- 135 high-level categories
- 7000 relations among them

Lexical
resources

Terminological
resources

Ontological
resources



Terminological resources

UMLS Metathesaurus

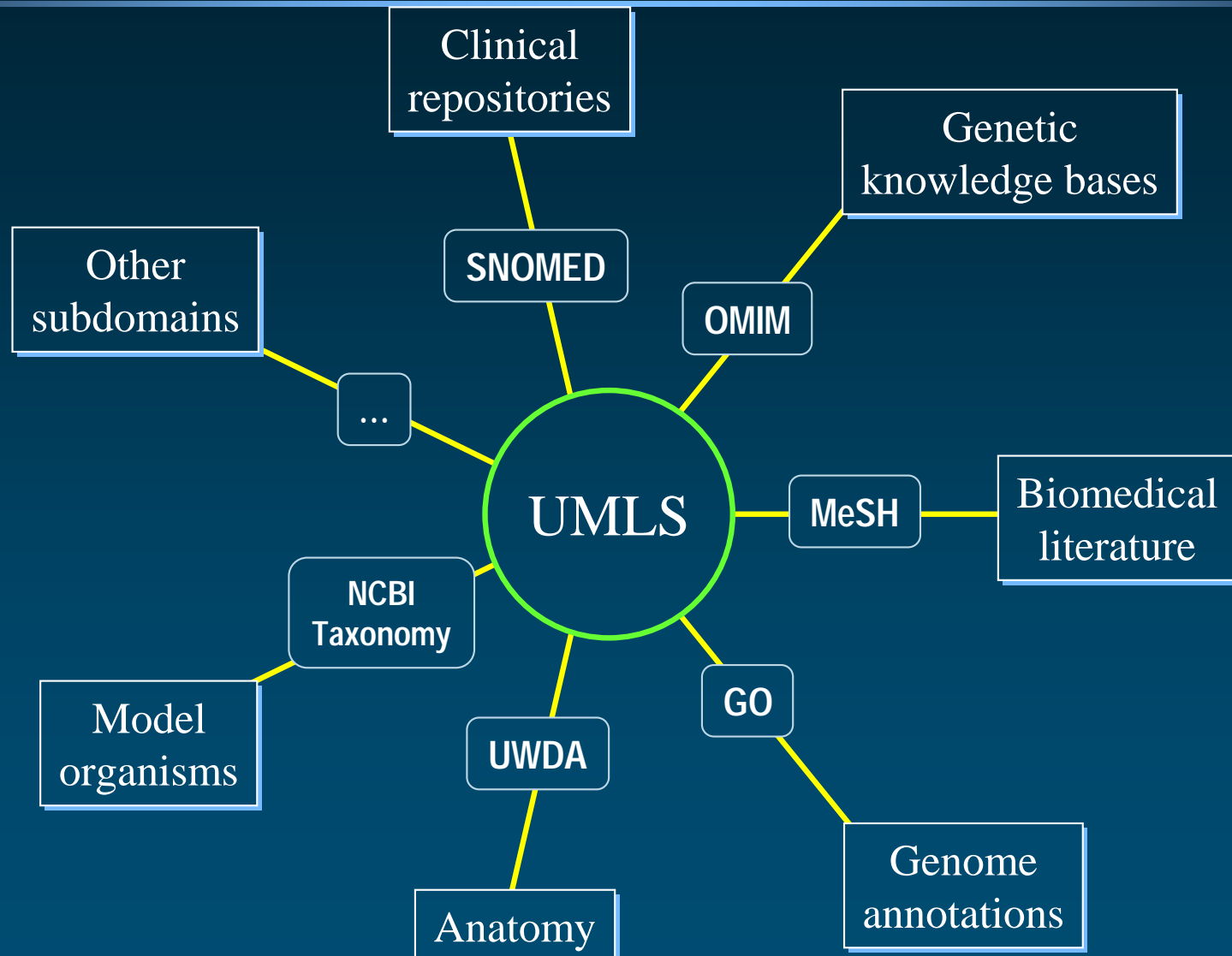
Source Vocabularies

(2005AB)

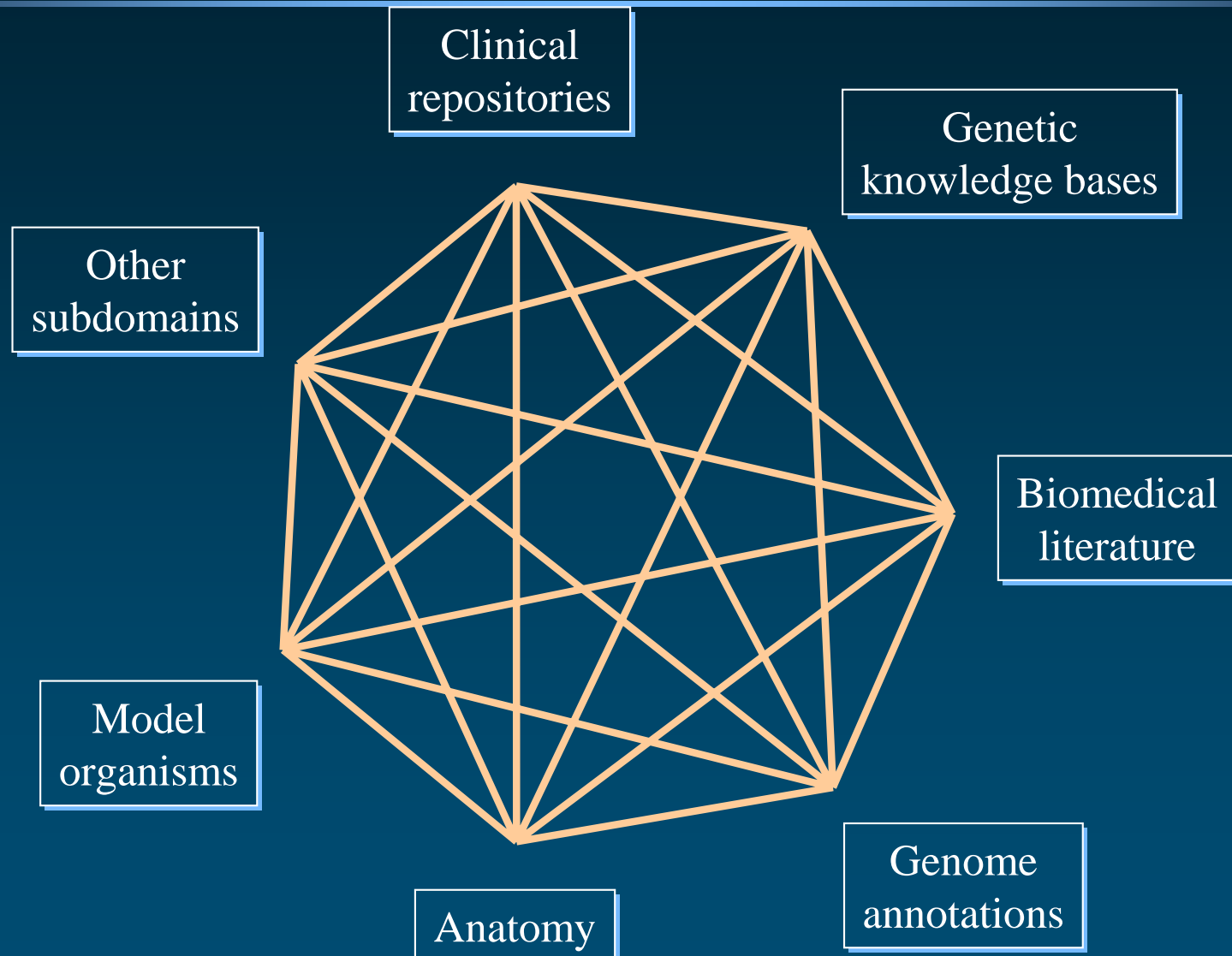
- ◆ 133 source vocabularies contributing concept names
- ◆ ~80 families of vocabularies
 - multiple translations (e.g., MeSH, ICPC, ICD-10)
 - variants (American-English equivalents, Australian extension/adaptation)
 - subsequent editions usually considered distinct families (ICD: 9-10; DSM: IIR-IV)
- ◆ Broad coverage of biomedicine
- ◆ Common presentation



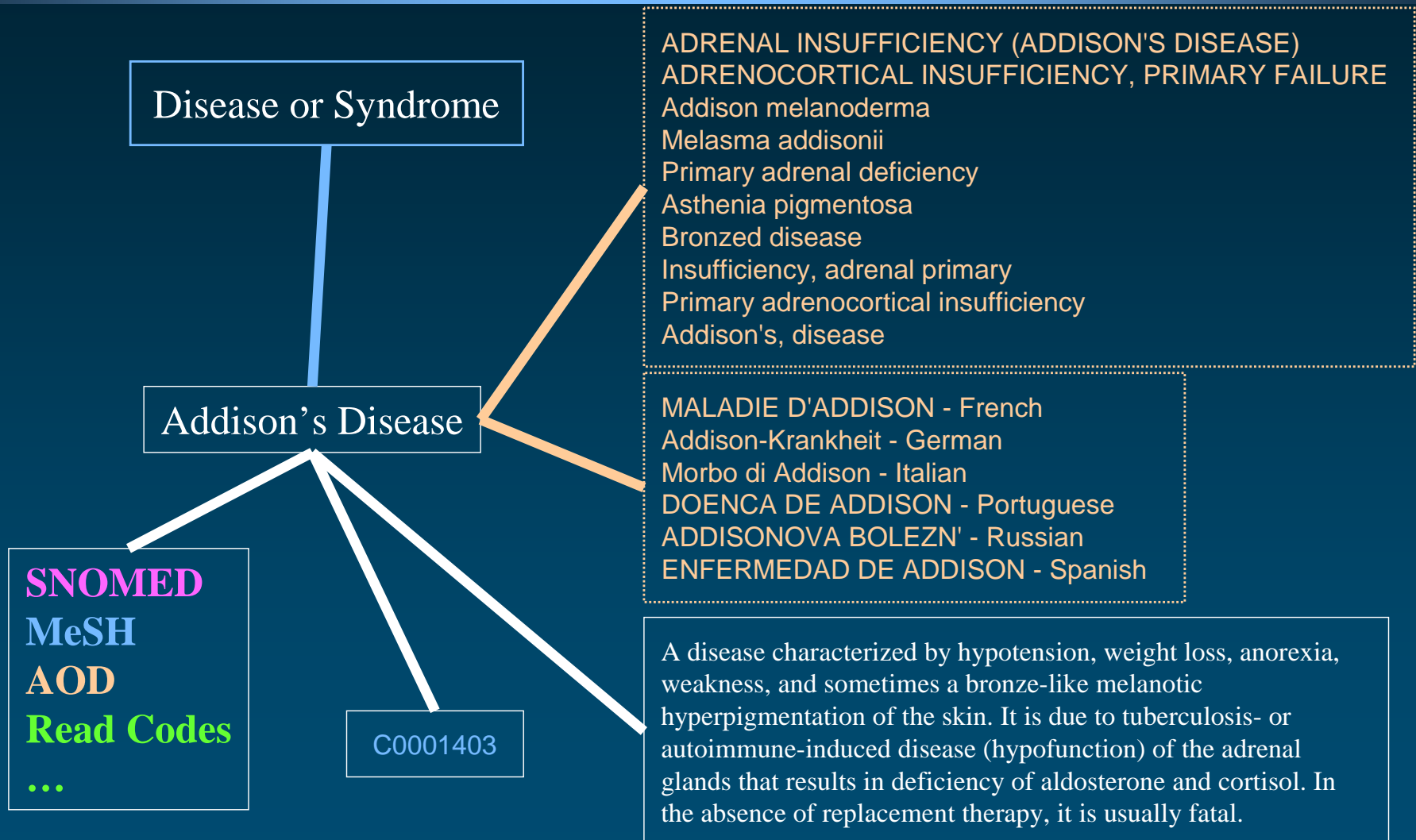
Integrating subdomains



Integrating subdomains

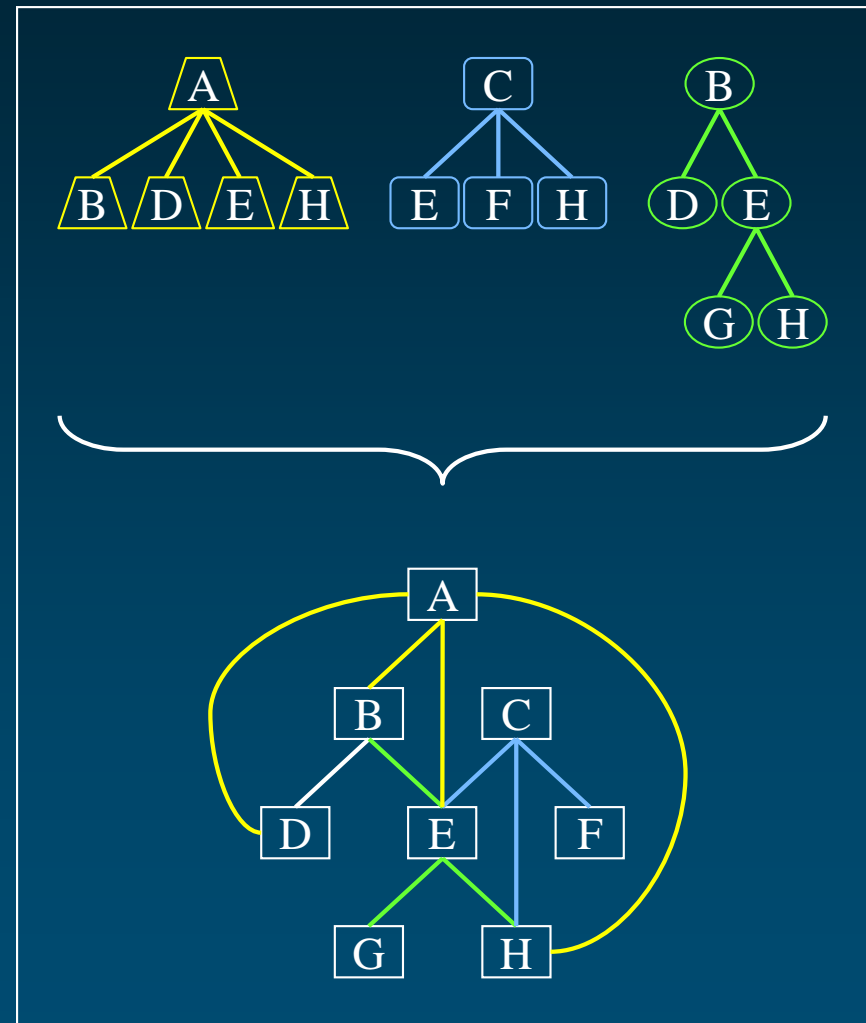


Addison's Disease: Concept



Organize concepts

- ◆ Inter-concept relationships: hierarchies from the source vocabularies
- ◆ Redundancy: multiple paths
- ◆ One **graph** instead of multiple **trees** (multiple inheritance)



Metaheasaurus relations Examples

◆ Neurofibromin 2

- Multiple parent concepts
 - Membrane proteins [MeSH]
 - Tumor suppressor proteins [MeSH]
 - Signaling protein [NCI Thesaurus]
- 1 child concept
 - Merlin, Drosophila [MeSH]
- Co-occurring concepts in MEDLINE
 - Neurofibromatosis 2 [13]
 - Membrane proteins [8]
 - ...



Finding Metathesaurus concepts in text

◆ MetaMap (MMTx)

- Developed at NLM
- Named entity recognition
- Approximate matches
- Used in many projects
- Not distributed with the UMLS

Neurofibromatosis type 2 (NF2) is often not recognised as a distinct entity from **peripheral neurofibromatosis**. **NF2** is a predominantly intracranial condition whose hallmark is bilateral **vestibular schwannomas**. **NF2** results from a **mutation** in the **gene** named **merlin**, located on **chromosome 22**.



Ontological resources

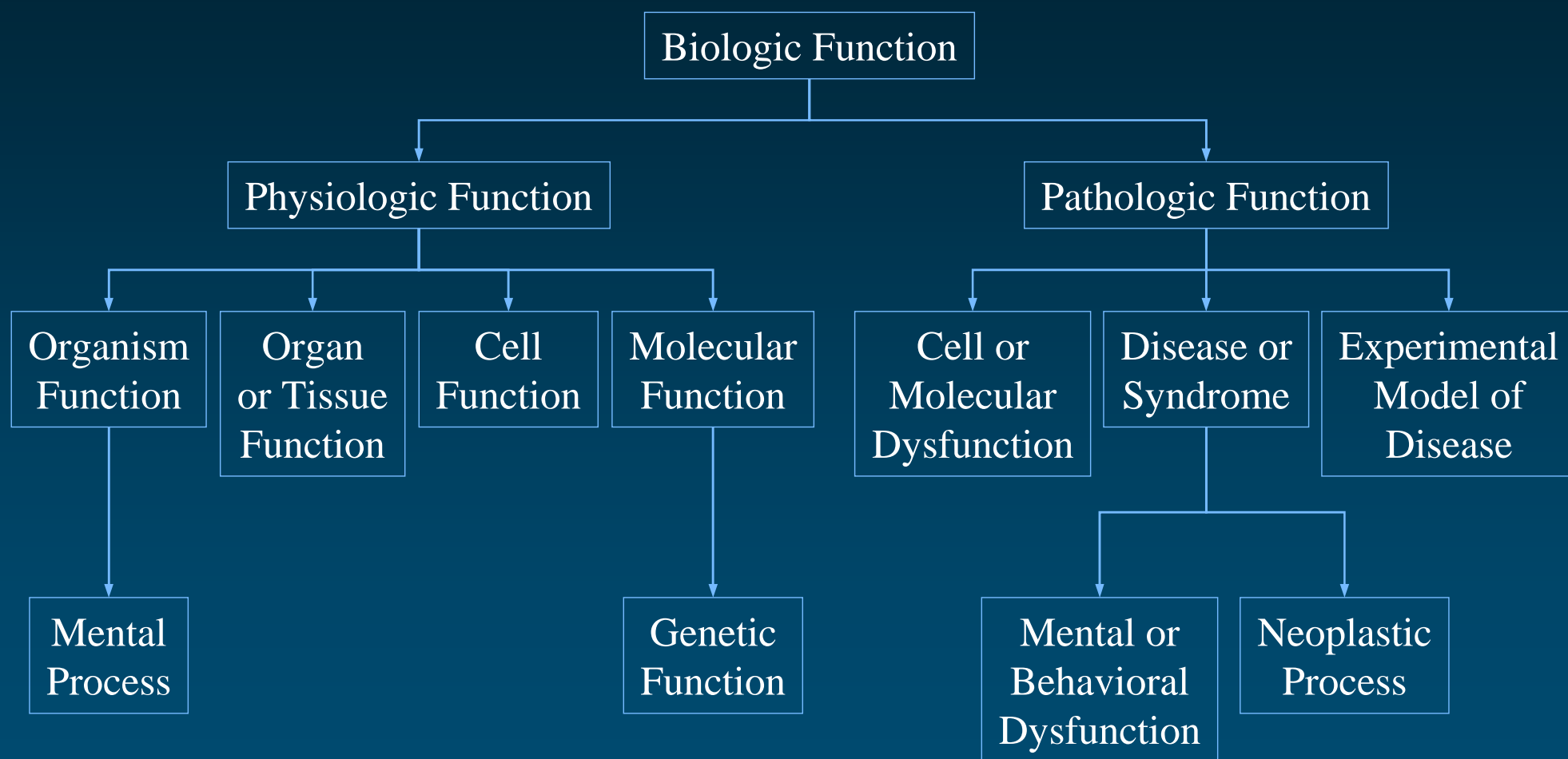
UMLS Semantic Network

Semantic Network

◆ Semantic types (135)

- tree structure
- 2 major hierarchies
 - Entity
 - Physical Object
 - Conceptual Entity
 - Event
 - Activity
 - Phenomenon or Process

“Biologic Function” hierarchy (isa)

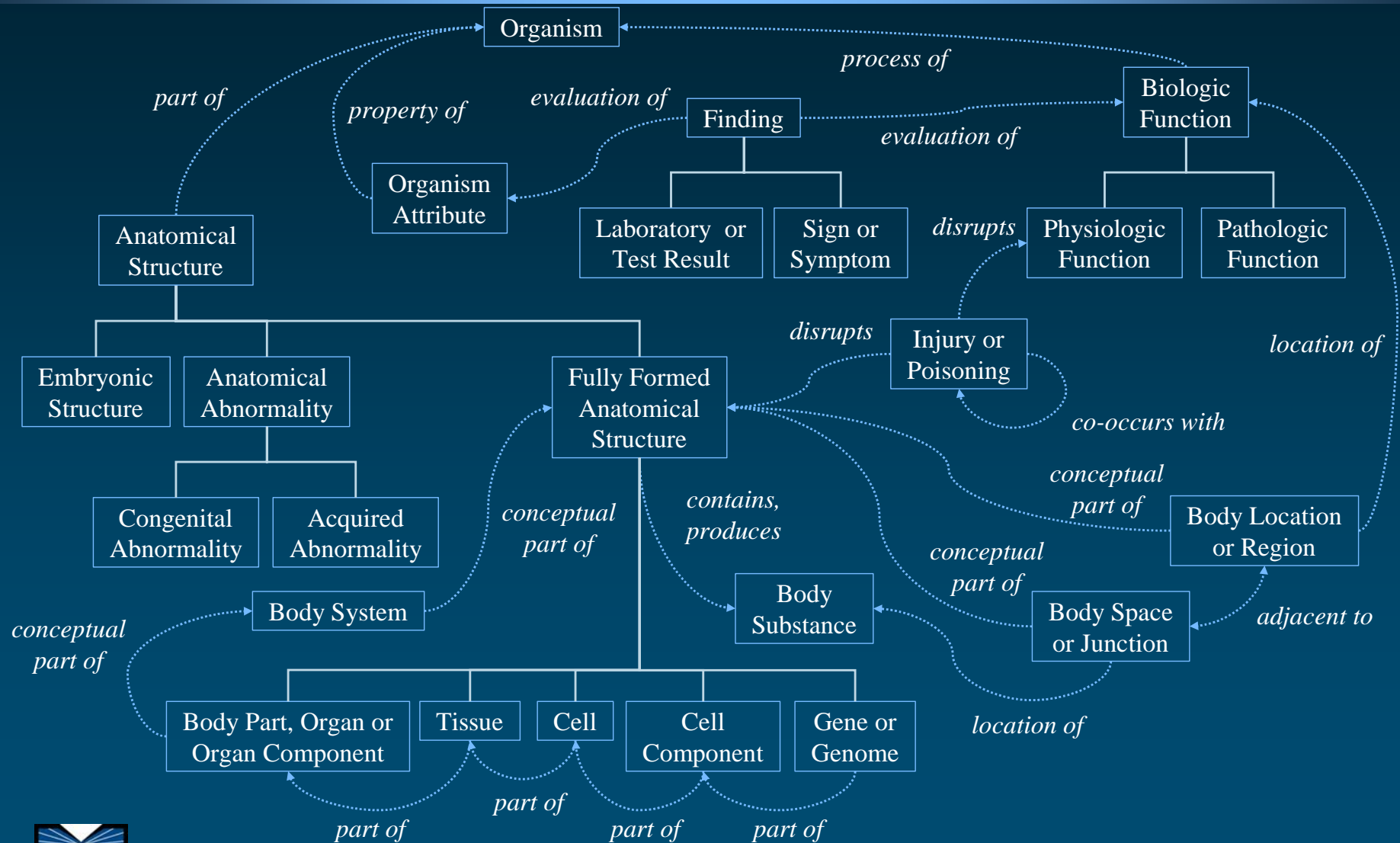


Semantic Network

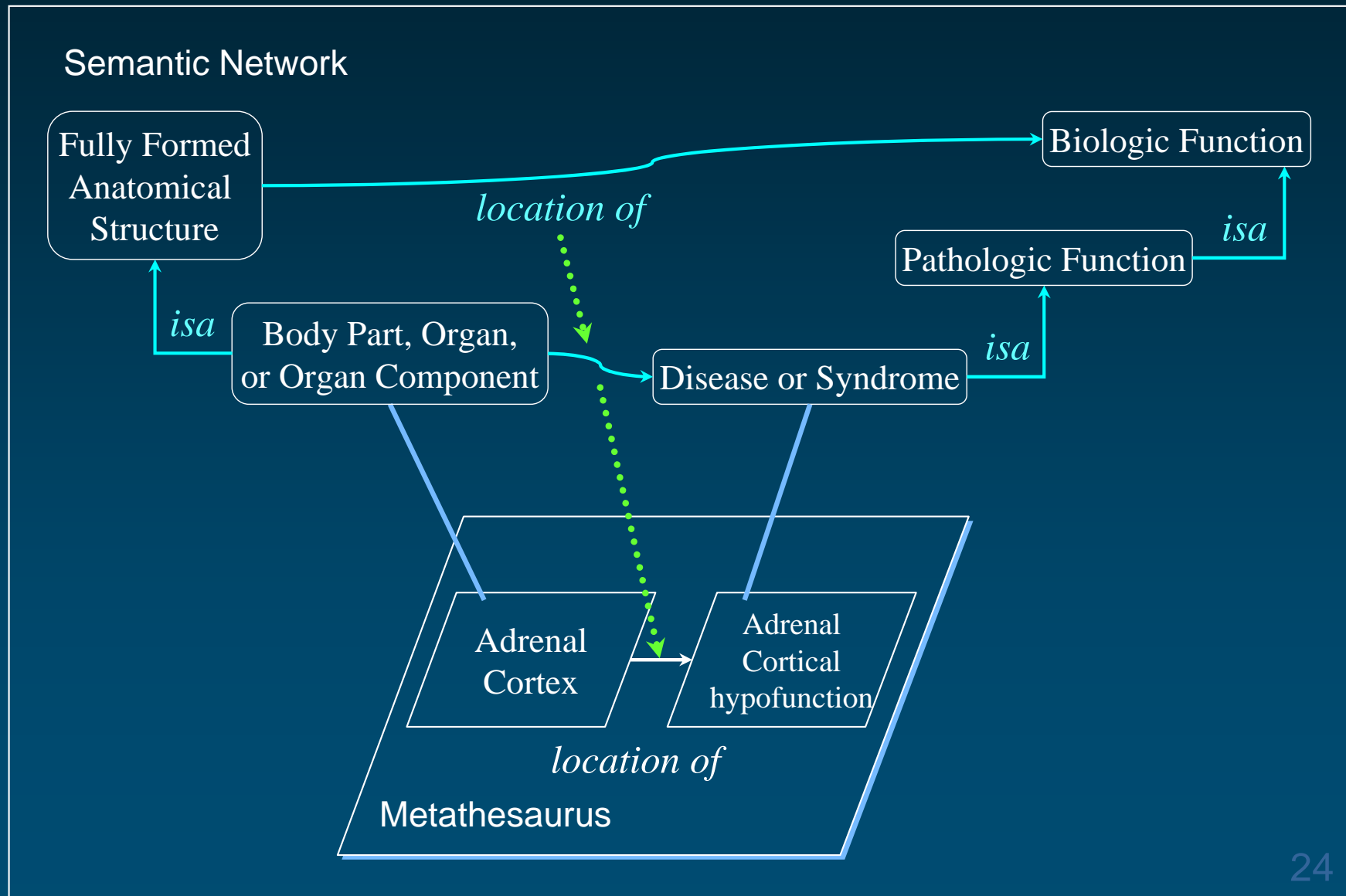
◆ Semantic network relationships (54)

- hierarchical (isa = is a kind of)
 - among types
 - **Animal** *isa* **Organism**
 - **Enzyme** *isa* **Biologically Active Substance**
 - among relations
 - *treats isa affects*
- non-hierarchical
 - **Sign or Symptom** *diagnoses* **Pathologic Function**
 - **Pharmacologic Substance** *treats* **Pathologic Function**

Associative (non-isa) relationships

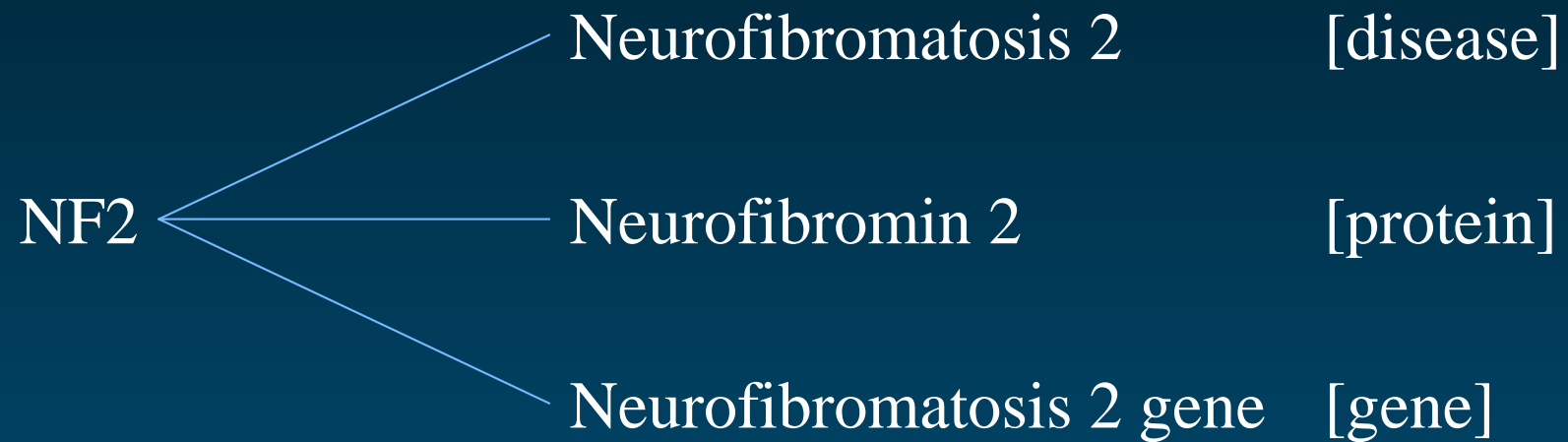


Relationships can inherit semantics



Some issues related to these resources

Ambiguity



Limited coverage

◆ e.g., Gene and protein names

- Additional sources
- Additional identification methods

Genew	http://www.gene.ucl.ac.uk/nomenclature/
Entrez Gene	http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=gene
UniProt	http://www.ebi.uniprot.org/index.shtml

Conclusions

Conclusions

- ◆ Lexical and terminological resources enable entity recognition
- ◆ Terminological and ontological resources enable relation extraction

But...

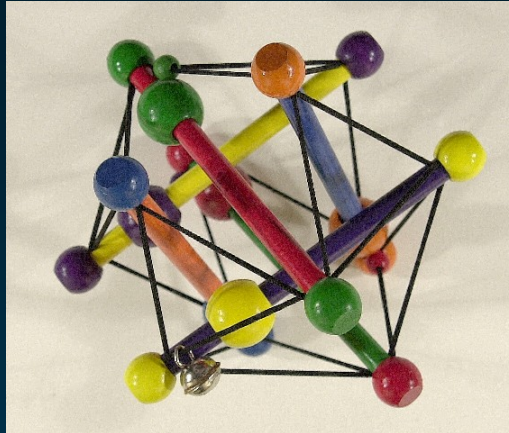
- ◆ Text mining techniques can also benefit
 - Terminologies: term extraction
 - Ontologies: ontology population



UMLS documentation and support

- ◆ UMLS homepage <http://umlsinfo.nlm.nih.gov/>
 - with links to all other UMLS information
- ◆ UMLSKS homepage <http://umlsks.nlm.nih.gov/>
 - with links to the User's and Developer's guides
- ◆ Email address for support custserv@nlm.nih.gov





Medical Ontology Research

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